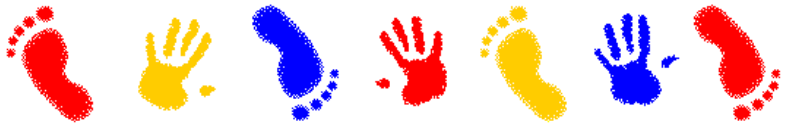
August 2007



Galactosemia

WHAT IS GALACTOSEMIA?

Galactosemia is a genetic disorder causing too much galactose in the blood. Galactose is a sugar. It may be found by itself, but is usually found as part of another sugar called lactose. Lactose, sometimes called milk sugar, is broken down in the body to galactose and glucose. Galactose is necessary as part of certain tissue components in the body. Excess galactose is normally changed into glucose in the liver. Glucose is the sugar used by the body for energy.

The baby with galactosemia cannot change galactose into glucose because the body is missing an enzyme todo this. If the galactosemic baby is untreated, galactose goes intot he blood and starts to collect. The blood then carries the galactose to different organs of the body, causing damage to the tissue.

NORMAL WAY OF MAKING ENERGY FROM LACTOSE IN THE BODY

Milk ⇒ Lactose ⇒ Galactose ⇒ Glucose ⇒ Energy

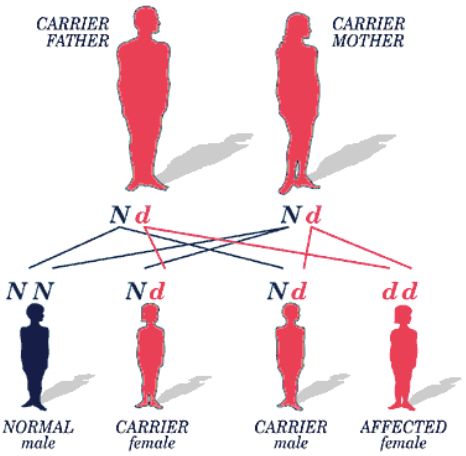
WHAT ARE THE SYMPTOMS OF GALACTOSEMIA?

The baby fails to grow and gain weight. The baby may cry frequently and have vomiting and stomach bloating. Symptoms begin to appear within days after milk feeding is started. Milk feeding is breast milk, most commercial formulas, or milk from cows or goats.

If the condition is not discovered in the first few days, other problems may develop from the build up of galactose. These include enlarged liver with cirrhosis, eye cataracts, infections, and mental retardation. Most of these problems can be reversed if galactosemia is discovered and treated early.

WHAT CAUSES GALACTOSEMIA?

Galactosemia is an inherited recessive genetic disorder. Both parents must carry a gene for galactosemia if an infant is to be affected. A carrier parent has one normal gene and one altered gene. The infant who has inherited the altered gene from each parent is affected since he or she is not able to convert galactose to glucose. The one normal gene in a carrier parent provides enough enzyme capability so this person does not show any signs of galactosemia. Thus people with an altered gene for the galactosemia enzyme rarely know that they are carriers before having an affected child.



N = normal gene

d = disease or abnormal gene

TESTING FOR GALACTOSEMIA

Galactosemia has been found to occur about once in 40,000 births. All infants born in Utah are screened for galactosemia at birth. Further testing is necessary to confirm the diagnosis. A sample of blood taken from the baby is tested for the enzyme level. A low level of enzyme may indicate galactosemia, a carrier of the galactosemia gene, or a variant of galactosemia. If too low, treatment should begin at once.

TREATMENT FOR GALACTOSEMIA

To treat galactosemia, all sources of galactose must be removed from the baby's diet. Most galactose in food is a part of lactose, the sugar found in milk, but other foods may contain related substances, which must also be avoided. The baby will be put on a soy-based milk substitute (ProSobee® or Isomil®) that does not contain lactose or galactose. Galactose and lactose are also found in many other foods and in some medicines. As a baby gets older, all dairy products and certain other foods will need to be restricted.

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Newborn Screening Program